Joint North South All Ireland Rare Disease Conference Abstract and Poster Booklet 2018

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Joint North South All Ireland Rare Disease Conference 2018

BOOK OF ABSTRACTS

Compiled by Julie McMullan & AJ McKnight

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Joint North South All Ireland Rare Disease Conference 2018:

Patients as Prime Movers in Rare Disease Research

Call for abstracts ***– Poster competition under the following categories:***

- Clinical / Laboratory investigations for Rare Diseases
- Epidemiology and Big Data contributing to Rare Disease Research
- Social Science considerations for Rare Diseases
- Improving Education for Rare Diseases

The Joint North South Rare Disease Conference 2018 will be held on 5th March in Riddel Hall, Queen's University, Belfast. The conference **offers participants the opportunity to be part of a global call on policy makers, researchers, companies and healthcare professionals to increasingly and more effectively involve patients in rare disease research.** The topic for the meeting is '*Nothing about us without us'* and aims to showcase examples of research; academic, clinical, and social, particularly where patients and families were engaged and involved; what difference it made; and what is happening now and into the future.

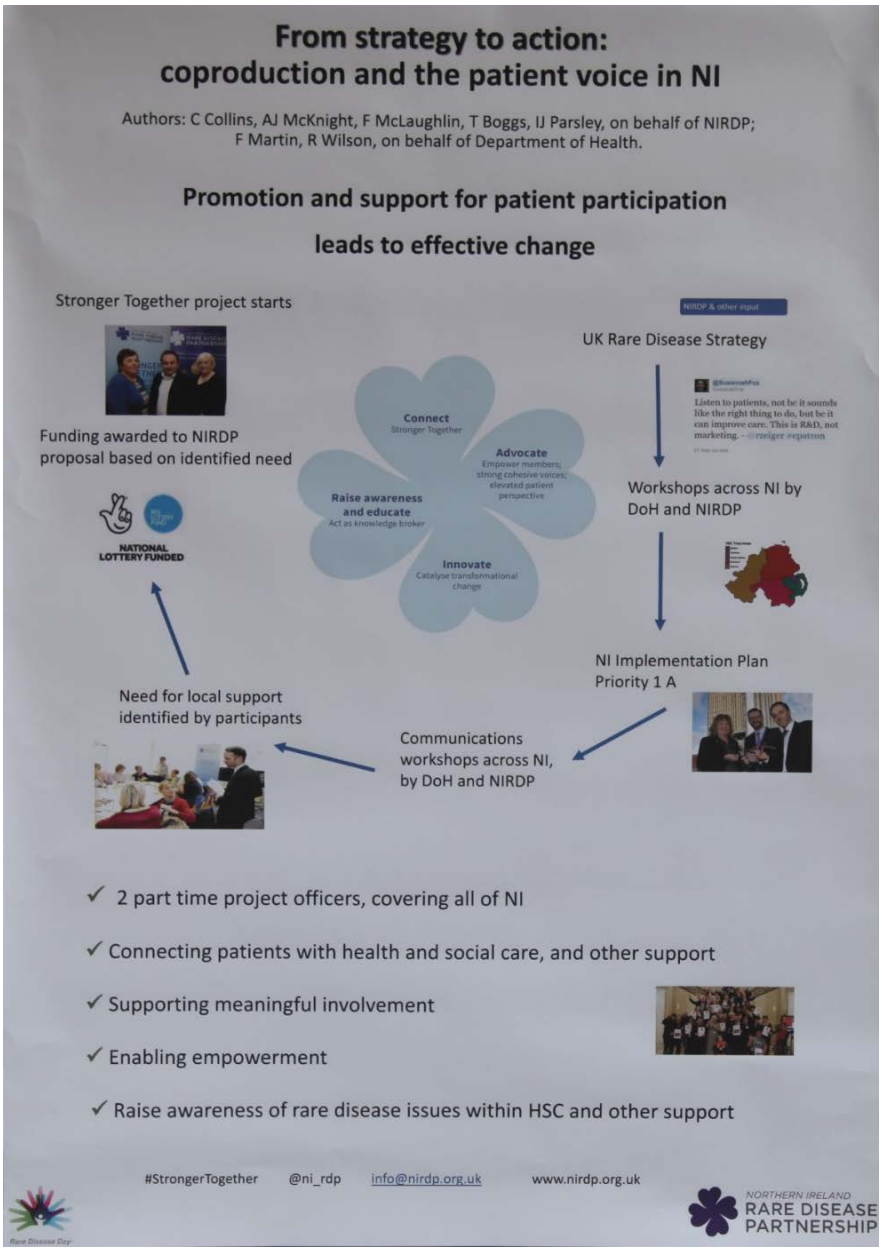
Abstracts (maximum 200 words, not including title and authors) should be emailed to j.mcmullen@qub.ac.uk, with 'POSTER COMPETITION' in the subject line, **by Monday 26th February 2018.** Posters must be A0 size in Portrait format and brought to Riddel Hall by 9.00am on 5 March 2018 for display.

A prize of £50 will be awarded to the winning entries in each category and a certificate given to each participant. CPD accreditation has been granted for this event.

From strategy to action: coproduction and the patient voice in NI:

Promotion and support for patient participation leads to effective change

Christine Collins, AJ McKnight, F McLaughlin, T Boggs, IJ Parsley on behalf of NIRDP;
F Martin, R Wilson on behalf of Department of Health



Improving Communication for Individuals with a Rare Condition

Ashleen Crowe, Amy Jayne McKnight and Helen McAneney

Centre for Public Health, Queen's University of Belfast.

A rare disease is defined as occurring in <1 in 2,000 people, but cumulatively rare diseases are common with one in 17 people in the UK being affected by a rare condition. The problems encountered because of the low number of instances of rare diseases are vast and need solutions. Thus improving communication mechanisms both within and around the healthcare system is of vital importance to individuals living and working with rare diseases.

Surveys amongst people affected by and working with a rare disease have been, and will continue to be, carried out to establish what communication for them is like within the healthcare system. The DELPHI model will then be used to identify the priorities for improving communication for people with rare disease in Northern Ireland.

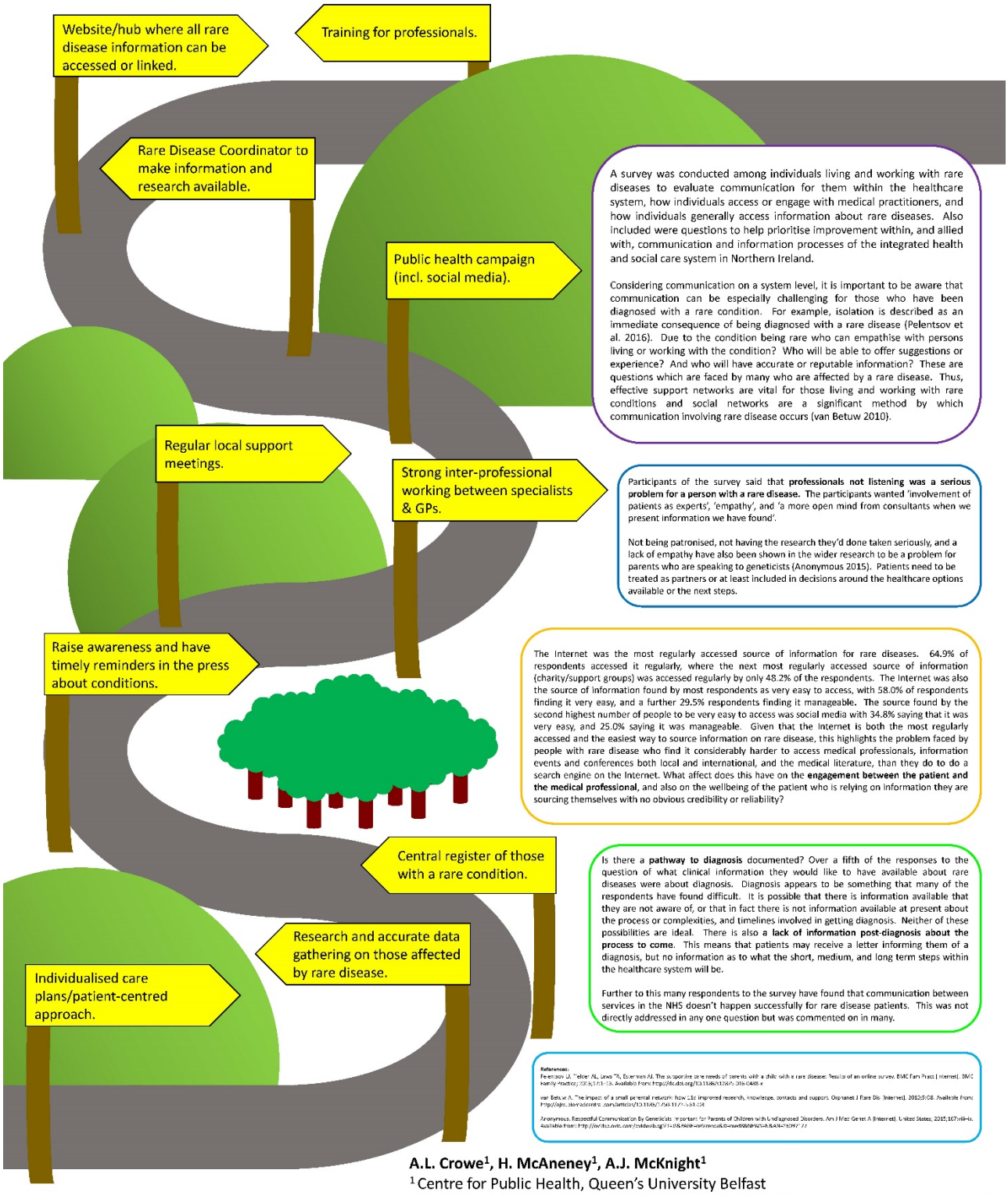
From the survey data there were 60 priorities identified which have been grouped under 4 headings: Sources of Information, Medical Care, Rare Disease Community, and Public Awareness.

There is a huge amount of progress which needs to be made in order to improve how someone with a rare disease receives information, treatment, and ultimately holistic care to help them deal with the life-changing event of discovering that they have a rare disease.

Signposts to change

by and for the rare disease community.

Each signpost directs to a source of information which people indicated as a priority for change within the Northern Ireland health and social care system.



A survey was conducted among individuals living and working with rare diseases to evaluate communication for them within the healthcare system, how individuals access or engage with medical practitioners, and how individuals generally access information about rare diseases. Also included were questions to help prioritise improvement within, and allied with, communication and information processes of the integrated health and social care system in Northern Ireland.

Considering communication on a system level, it is important to be aware that communication can be especially challenging for those who have been diagnosed with a rare condition. For example, isolation is described as an immediate consequence of being diagnosed with a rare disease (Pelentsov et al. 2016). Due to the condition being rare who can empathise with persons living or working with the condition? Who will be able to offer suggestions or experience? And who will have accurate or reputable information? These are questions which are faced by many who are affected by a rare disease. Thus, effective support networks are vital for those living and working with rare conditions and social networks are a significant method by which communication involving rare disease occurs (van Betuw 2010).

Participants of the survey said that **professionals not listening was a serious problem for a person with a rare disease.** The participants wanted 'involvement of patients as experts', 'empathy', and 'a more open mind from consultants when we present information we have found'.

Not being patronised, not having the research they'd done taken seriously, and a lack of empathy have also been shown in the wider research to be a problem for parents who are speaking to geneticists (Anonymous 2015). Patients need to be treated as partners or at least included in decisions around the healthcare options available or the next steps.

The Internet was the most regularly accessed source of information for rare diseases. 64.9% of respondents accessed it regularly, where the next most regularly accessed source of information (charity/support groups) was accessed regularly by only 48.2% of the respondents. The Internet was also the source of information found by most respondents as very easy to access, with 58.0% of respondents finding it very easy, and a further 29.5% respondents finding it manageable. The source found by the second highest number of people to be very easy to access was social media with 34.8% saying that it was very easy, and 25.0% saying it was manageable. Given that the Internet is both the most regularly accessed and the easiest way to source information on rare disease, this highlights the problem faced by people with rare disease who find it considerably harder to access medical professionals, information events and conferences both local and international, and the medical literature, than they do to do a search engine on the Internet. What affect does this have on the **engagement between the patient and the medical professional**, and also on the wellbeing of the patient who is relying on information they are sourcing themselves with no obvious credibility or reliability?

Is there a **pathway to diagnosis** documented? Over a **fifth** of the responses to the question of what clinical information they would like to have available about rare diseases were about diagnosis. Diagnosis appears to be something that many of the respondents have found difficult. It is possible that there is information available that they are not aware of, or that in fact there is not information available at present about the process or complexities, and timelines involved in getting diagnosis. Neither of these possibilities are ideal. There is also a **lack of information post-diagnosis about the process to come.** This means that patients may receive a letter informing them of a diagnosis, but no information as to what the short, medium, and long term steps within the healthcare system will be.

Further to this many respondents to the survey have found that communication between services in the NHS doesn't happen successfully for rare disease patients. This was not directly addressed in any one question but was commented on in many.

References:

Pelentsov LJ, Fielder AL, Laws TA, Esterman AJ. The supportive care needs of parents with a child with a rare disease: Results of an online survey. BMC Fam Pract [Internet]. BMC Family Practice; 2016;17:1–13. Available from: http://dx.doi.org/10.1186/s12875-016-0488-x

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Anonymous. Respectful Communication By Geneticists Important for Parents of Children with Undiagnosed Disorders. Am J Med Genet A [Internet]. United States; 2015;167:viii–ix. Available from: http://ovidsp.ovid.com/ovidweb.cgi?T=JS&PAGE=reference&D=med8&NEWS=N&AN=25997777

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Improving communication for young people managing a medical condition: Evaluation of 'My Healthcare Passport'

Dr Janet Diffin and Dr Peter O'Halloran

The School of Nursing and Midwifery, Queen's University Belfast

Most professionals agree that young people with life-threatening or life-limiting conditions, and their families, should be fully involved in making decisions related to their care. However, fragmented health and social services can often make communication challenging. Young people often have to repeat their story to different professionals. The use of a patient-held record (PHR) might help overcome such communication difficulties. 'My Healthcare Passport' is a PHR for people facing long-term illness (developed through consultation with 41 stakeholder groups throughout Northern Ireland). It helps people to share important information between their families, carers, and healthcare professionals. Our research project involves working with young people aged 16 – 24 years old with a medical condition to evaluate the usefulness of 'My Healthcare Passport' (this include certain rare diseases). We will ask them to use the healthcare passport for up to nine months and speak with them three times to find out how useful it has been for them, and whether any improvements are needed. The results will provide valuable insight into how 'My Healthcare Passport' could be used successfully in the future, and reveal the potential benefits of it to young people managing a medical condition, including those with rare diseases.

Improving communication for young people manging a medical condition: Evaluation of 'My Healthcare Passport'

Dr Janet Diffin & Dr Peter O'Halloran

QUEEN'S UNIVERSITY BELFAST

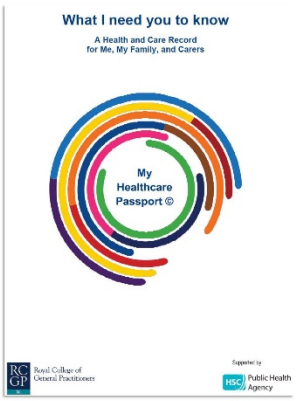
Background

- Communication with service providers can be challenging for young people managing a medical condition. Young people have reported having to repeat their story to different professionals.
- The use of a patient-held record, such as **'My Healthcare Passport'** might help overcome such communication difficulties.

My Healthcare Passport is designed to be completed, updated and kept by the patient, or any member of their family or carers.

It acts as a core record of how their health is evolving and of information required to support their health and wellbeing.

My Healthcare Passport



- All About Me
- Information/ Advice from Healthcare
- I Need Help With
- Issues of Urgency
- My Health
- My Medical Condition
- My Medication
- My Support Team
- My Healthcare Team
- Useful Resources
- In the Event of...

Study Aims

- To find out how useful 'My Healthcare Passport' is to young people aged **16 – 24 years old** with a complex medical condition, and their families.
- To identify what factors are needed to make the use of a healthcare passport a success.

We will ask the young person to:

- ✓ use 'My Healthcare Passport' for **up to nine months.**
- ✓ Speak with us on three occasions.
- ✓ keep a diary about their experiences of using the passport.
- ✓ let us see how they have used their passport (with their permission).

Study progress and initial feedback

- **Nine** young people and/or parents recruited.

> "it would've been very good if they had it at the very start, when they were first diagnosed and then you could've kept quite a lot of details, this could've been very useful".

> "when you go to clinics or, I think that they always often ask what medicines they're on and you can have a dosage in the medicines and everything, and just get to say this is what they are sort of thing".

Conclusion

- This study will provide valuable insight into how 'My Healthcare Passport' could be used successfully in the future, and reveal the potential benefits of it to young people managing a complex medical condition, including those with rare diseases.

Contact details for further information
Dr Janet Diffin | Queen's University Belfast | **Email:** j.diffin@qub.ac.uk
Twitter: @HCPassportStudy
Website: thepassportstudy.wordpress.com

This study was funded by Marie Curie

Marie Curie Care and support through terminal illness

Opsoclonus Myoclonus Ataxia Syndrome: Review of long term outcomes

Nichola Ejaz

Dancing Eye Syndrome Support Trust

Aim: Opsoclonus Myoclonus Ataxia Syndrome (OMAS), also known as Dancing Eye Syndrome (DES), is a rare inflammatory neurological condition which is thought to be due to an autoimmune process. Onset of OMAS/DES often has paraneoplastic or post-infectious origins. However, a percentage of cases are classified as idiopathic. The tumour type is commonly a neuroblastoma. Treatment of the neuroblastoma, while important in its own right, does not appear to alter the outcome of Opsoclonus Myoclonus Ataxia Syndrome. Long term neurological and cognitive sequelae are often reported.

Methodology: A review of literature, questionnaires and surveys.

Results: Results demonstrate that a large proportion of childhood onset OMAS/DES patients experience ongoing neurological sequelae and an episodic nature of disease progression.

Implications: Patients with OMAS/DES have represented reduced connectivity and grey matter in fMRI imaging which may be representative of the long term outcome of these patients. Hyperkinetic movement disorders, ataxia, learning disability, anxiety, obsessive-compulsive disorder and cognitive decline are examples of long term sequelae. Patients often report an episodic nature of this syndrome due to certain triggers such as infection. The onset of puberty seems to be a critical window, the transition through which seems to reflect and also predict whether the disease course is monophasic or episodic and progressive in nature.

OPSOCLONUS MYOCLONUS ATAXIA SYNDROME: REVIEW OF LONG TERM OUTCOMES

Nichola Ejaz

Dancing Eye Syndrome Support Trust (UK) and OMSLife (US)

ABSTRACT

Reason for writing: Opsoclonus Myoclonus Ataxia Syndrome (OMAS), also known as Dancing Eye Syndrome (DES), is a rare inflammatory neurological condition which is thought to be due to an autoimmune process. Onset of OMAS/DES often has paraneoplastic or post-infectious origins. However, a percentage of cases are classified as idiopathic. The tumour type is commonly a neuroblastoma. Treatment of the neuroblastoma, while important in its own right, does not appear to alter the outcome of Opsoclonus Myoclonus Ataxia Syndrome. Long term neurological and cognitive sequelae are often reported.

Problem: Long term outcomes of childhood onset OMAS/DES are poorly understood and proves to be a massive hurdle for many of the patients. Long term neurological and cognitive sequelae are often reported.

Methodology: A review of literature, questionnaires and surveys.

Results: Results demonstrate that a large proportion of childhood onset OMAS/DES patients experience ongoing neurological sequelae and an episodic nature of disease progression.

Implications: Patients with OMAS/DES have represented reduced connectivity and grey matter in fMRI imaging at a whole brain level which may be representative of the long term outcome of these patients. Hyperkinetic movement disorders, ataxia, learning disability, anxiety, obsessive-compulsive disorder and cognitive decline are examples of long term sequelae. Patients often report an episodic nature of this syndrome due to certain triggers such as infection. The onset of puberty seems to be a critical window, the transition through which seems to reflect and also predict whether the disease course is monophasic or episodic and progressive in nature.

BACKGROUND

Opsoclonus Myoclonus Ataxia Syndrome (OMAS), also known as Dancing Eye Syndrome (DES), or Kinsbourne Syndrome, is a rare inflammatory neurological condition which is often thought to be due to an autoimmune process. Onset of OMAS/DES often has paraneoplastic or post-infectious origins, however a percentage of cases are classified as idiopathic. The tumour type most commonly associated with OMAS/DES is neuroblastoma. The key features are unsteadiness (ataxia), jerky movements of the trunk and limbs (myoclonus), rapid involuntary eye movements in all directions (opsoclonus) and usually marked irritability with sleep disturbance. The occurrence of marked irritability with behavioural change and sleep disturbance in an infant with new onset ataxia is one of the strongest clues that this may be OMAS/DES. Muscle tone seems to decrease causing floppiness and lethargy. Speech difficulties also occur, for instance losing previously-fluent speech or not speaking at all. Most children develop the condition in the second or third year of life but sometimes can occur earlier or later. Occurrence in adulthood can also occur. Most children identified with a neuroblastoma will need to undergo staging and surgical resection, followed by monitoring by the neuro-oncology team.
Treatment of the neuroblastoma, while important in its own right, does not appear to alter the outcome of Opsoclonus Myoclonus Ataxia Syndrome. Long term outcomes of OMAS/DES are poorly understood and proves to be a massive hurdle for many of our patients. Current treatment protocols promote a combination of therapies including immunoglobulins, monoclonal antibodies and steroids. Plasmapheresis has occasionally been used with some success in non paraneoplastic cases.

CURRENT TREATMENT SCHEDULE

The aims of treatment are firstly to treat the neuroblastoma tumour if present and secondly to dampen down the immune system to reduce the attacks to the cerebellum and prevent any lasting damage. The immune system is modulated by using a combination of medicines. A triple therapy has proven most effective for OMAS/DES onset using steroids, monoclonal antibodies and immunoglobulins. Treatment continues until the symptoms of OMAS/DES improve.

This triple therapy appears to result in a better cognitive outcome in conjunction with a rapid diagnosis so treatment can be provided promptly.

However, a treatment plan is not available for the long term sequelae that appears to occur where the OMAS/DES has not achieved a monophasic course.

METHODOLOGY

To better understand the long term outlook of this condition, a review of literature, questionnaires and surveys associated with both the Dancing Eye Syndrome Support Trust (UK) and OMSLife (US) has been made to identify the long term outcomes of Opsoclonus Myoclonus Ataxia Syndrome into adulthood.

RESULTS

1) Questionnaire: demonstrated that 91% of childhood-onset OMAS/DES patients felt that they had long term problems resulted from this condition (MaCleod, 2014).

QUESTIONNAIRE 1 (MaCleod, 2014):

The aims of the questionnaire were:
1) **To raise awareness that OMS/DES exists beyond childhood**
2) **2) To give a voice and platform to these young people**
3) **3) To highlight the lifelong support required to help these young people**

The questions were both closed and open and covered diagnosis, symptoms, relapses, education, family life, relationships, leisure activities and employment. There were 23 respondents, a response rate of approximately 43%. 20 females and 3 males, with an age range from 13-30 years. Some responses supported other studies eg majority of children were aged <2 years at diagnosis and approx 56% had had a neuroblastoma

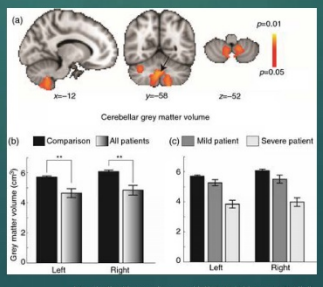
However, the questionnaire also highlighted a number of other areas:
- **91% have suffered one or more relapses-with detailed descriptions of individual's symptoms**
- **Nearly ALL respondents STILL have symptoms to varying degrees**
- **77% of school leavers went on to further education but many didn't complete their courses and felt OMS was a major factor in this**
- **Only 33% of those eligible to work were in paid employment with 22% working as volunteers**

http://www.dancingeyes.org.uk/downloads/Report%20on%20OMS%202014%20workshop%207.pdf

2) Literature: Following this report an imaging study was completed which determined cerebellar grey matter loss in teenagers and young adults who had a historical diagnosis of childhood onset OMAS/DES (Geetha, 2015)

RESEARCH ARTICLE (Anand et al, 2014):

i.



http://onlinelibrary.wiley.com/doi/10.1111/dmcn.12594/full

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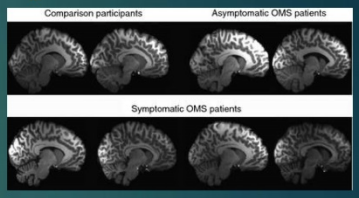
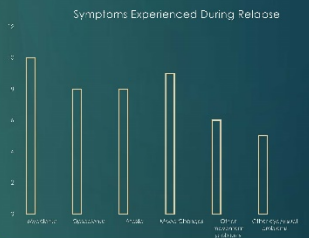


3) Survey: To clarify symptoms of relapse, eleven patients regarding instances of relapse from teenage years and beyond was obtained the following (Ejaz, 2018).



Other movement problems declared: tremor, dystonia, restless legs and loss of mobility.

Other eye/visual problems declared: photosensitivity, loss of vision, blurry vision, nystagmus, strabismus, oscillopsia, double vision, visual snow, loss of depth perception and spatial awareness.

Tinnitus is also reported.

Causes of relapse reported included: puberty, pregnancy, postpartum, lactation, infection, medications, HRT and stress.

DISCUSSION

Questionnaires and surveys have identified key features regarding the nature of long term outcomes experienced. Patients with OMAS/DES have represented reduced connectivity and grey matter in fMRI imaging at a whole brain level which may be representative of the long term outcome of these patients (Anand et al, 2014). The episodic nature of OMAS/DES is still not understood and is poorly documented in literature. Hyperkinetic movement disorders, ataxia, learning disability, anxiety, obsessive-compulsive disorder and cognitive decline are examples of long term sequelae. Patients often report an episodic nature of this syndrome due to a worsening or reoccurrence of symptoms. Reported triggers have included infection, contraindicated medications, vaccinations, puberty, pregnancy and possibly food intolerances to gluten. The onset of puberty seems to be a critical window, the transition through which seems to reflect and also predict whether the disease course is monophasic or episodic and progressive in nature. Larger studies would be beneficial.

REFERENCES

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- Morag MaCleod / Dancing Eye Syndrome Support Trust (2014) *Reporting long term outcomes of OMS*. Dancing Eye Syndrome Support Trust
- Nichola Ejaz / Dancing Eye Syndrome Support Trust (2018) *Symptoms experienced during relapse*, Survey on OMS Warriors

The measurement of rare cancers

Anna Gavin, Colin Fox, Ronan Campbell, Sinead Lardner, Eileen Morgan

N. Ireland Cancer Registry, Queens University, Belfast

The N. Ireland Cancer Registry founded in 1994 provides information on cancers and pre-malignant disease occurring in Northern Ireland for research, education, service monitoring and planning. It collects morphology codes using international classifications and has data on pathologically rare tumours which arise in non-rare anatomical sites. E.g. Anaplastic carcinoma of the thyroid.

The registry provides a secure confidential environment for the storage and analysis of the sensitive clinical data having received ISO 27001 accreditation. It has links with the NI biobank facilitating the collection of clinical details on biological samples

From 2011 to 2015, on average, there were 4,557 male and 4,516 female patients diagnosed with cancer each year excluding Non-Melanoma Skin Cancer (NMSC). There were an additional 2,065 male and 1,576 female patients diagnosed with NMSC. Included in this total are many rare cancers usually categorised as 'other' amounting to approximately 1200 cases per year

Work is ongoing at European and international level to standardise recording and categorisation of these rare tumours and to combine datasets from many countries to enable epidemiological and scientific study including Joint Action on Rare cancers work package 4 - Improving epidemiological surveillance on rare cancers in Europe.

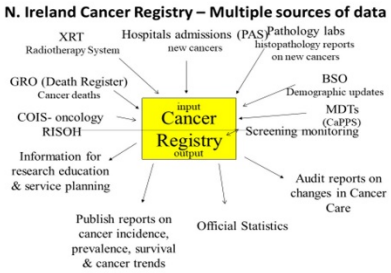
THE MEASUREMENTS OF RARE CANCERS

Anna Gavin, Colin Fox. Sinead Lardener, Eileen Morgan
Northern Ireland Cancer Registry. Queens University Belfast

Introduction

The N. Ireland Cancer Registry founded in 1994 provides information on cancers and pre-malignant disease occurring in Northern Ireland for research, education, service monitoring and planning. It collects morphology codes using international classifications and has data on pathologically diagnosed rare tumours which arise in non-rare anatomical sites. E.g. Anaplastic carcinoma of the thyroid.

NICR Methods of working



The registry provides a secure confidential environment for the storage and analysis of the sensitive clinical data having received ISO 27001 accreditation. It has links with the NI biobank facilitating the collection of clinical details on biological samples

Statistics

From 2011 to 2015, on average, there were 4,557 male and 4,516 female patients diagnosed with cancer each year excluding Non-Melanoma Skin Cancer (NMSC). There were an additional 2,065 male and 1,576 female patients diagnosed with NMSC. Included in this total are many rare cancers usually categorised as 'other' amounting to approximately 1200 cases per year , some examples of these , their codes and number of cases is below

Site	ICD 10 code	Morphology	Average number per year*
Hepatocellular carcinoma of liver and intrahepatic bile tract (IBT)	C22	8170-8175, 8180	172
Chronic myeloid leukaemia	C92.1, C92.2	9863, 9875	15
Mixed epithelial and mesenchymal tumours of the uterus	C53.0-C55.9	8933,8950-8951, 8980	13
Epithelial tumours of nasal cavity and sinuses	C30.0-C31	8000, 8001, 8004, 8010, 8011, 8020-8022, 8032, 8050-8076, 80878, 8082-8084, 8123, 8144, 8560, 8980	8
Gastrointestional stromal sarcoma	Any	8936	5
Malignant melanoma of uvea	C69.3-C69.4	8720-8780	3
Kaposi's sarcoma	Any	9140	2
Malignant meningiomas	C70	9530, 9538-9539	1

Work is ongoing at European and international level to standardise recording and categorisation of these rare tumours and to combine datasets from many countries to enable epidemiological and scientific study including Joint Action on Rare cancers work package 4 - Improving epidemiological surveillance on rare cancers in Europe.

The N. Ireland Cancer Registry is funded by the Public Health Agency for Northern Ireland.

Email: nicr@qub.ac.uk Tel: +44 (0)28 9063 2573 Website: www.qub.ac.uk/nicr

Genomic and Bioinformatic interrogation of rare diseases: Translating multi-omic research into clinical progress.

Katie Kerr, Helen McAneney, AJ McKnight

Centre for Public Health, Queen's University Belfast

The paradox of rare diseases is that they are individually rare, yet cumulatively over 350 million people are affected by rare diseases worldwide. There are approximately 8000 rare disorders, very often with variable clinical presentations, which makes diagnosing rare diseases challenging. Two in five rare disease patients report difficulties obtaining diagnosis, with many patients waiting several years between first symptoms and diagnosis. Without diagnosis patients report decreased quality of life, negative impacts on mental health, poor prognosis and difficulties in accessing an effective treatment plan. Therefore recent research has been focused on improving diagnosis, including molecular characterisation of a rare disease using multi-omic approaches. This evaluates differences at the genomic sequence level, such as whole genome sequencing through the 100,000 genomes project, but also examining epigenomic changes such as differential methylation.

This project reviews current research into multi-omics of rare disease and ultimately leverages multi-omic analyses to identify diagnostic biomarkers where whole genome sequencing has been insufficient to render a diagnosis. These allied approaches to improve rare disease diagnosis will benefit people living and working with rare disease and provide insight into the biological mechanisms of disease leading to potentially novel therapeutics.

A multi-omic approach to diagnosing rare disease

Katie Kerr, Dr Helen McAneney and Dr AJ McKnight.

INTRODUCING MULTI-OMICS

- Multi-omics is an approach to biological research which looks at multiple 'omes' including the **genomics**, **epigenomics** and **transcriptomics.**

(Check out the glossary below if you're not sure what any terms mean!)

- The 100,000 genomes project[2] is a UK based project which is attempting **whole genome sequencing** of patients with rare disease and their families to aid in diagnosis.
- From this data we can then carry out multi-omic research, increasing the likelihood of obtaining a diagnosis, which leads us to this project's research...

THIS RESEARCH PROJECT INVOLVES...

- Conducting **systematic reviews** into the current literature surrounding multi-omics and rare disease.
- Epigenomic and transcriptomic analyses of patient samples from the 100,000 genomes project which will have been subjected to whole genome sequencing but it was insufficient to render a diagnosis.
- Specifically I will be studying differential **methylation** (epigenomics) and **gene expression**, or **RNA levels** (transcriptomics).

PATIENT CENTERED RESEARCH

- The paradox of rare disease is that whilst they are individually rare, approximately **350 million people worldwide**[1] are affected...
- There are an estimated **8000 types of rare disorders**[1], often with variations in clinical presentation, making diagnosis challenging...
- **Patients can wait several years for a diagnosis**, which can negatively impact prognosis, quality of life and make access to effective treatment and support difficult!

So what are researchers doing to help?

HOPE FOR THE FUTURE

By utilising an allied multi-omic approach to studying rare disease we will be able to...

- Improve diagnosis speed and accuracy.
- Improve our understanding of the biological mechanisms behind rare disease.
- Identify potentially novel therapeutics.

Therefore, multi-omic approaches to studying rare disease will positively impact the lives of people living and working with rare disease.

Watch me!

Here's lots of patient experiences which give insights into living with a rare disease, including difficulties in obtaining a diagnosis.

Glossary of genetics terms

- **Genomics**: The study of the structure, function and evolution of a persons genetic material.
- **Epigenomics**: The study of non-sequence level DNA modifications which effects gene activity.
- **Methylation**: The act of adding a chemical methyl group which can effect gene activity levels.
- **RNA**: Ribonucleic acid which does many things, including acting as a messenger (mRNA) that carries instructions from the DNA to the cells to carry out cell duties, like synthesise proteins!
- **Gene expression**: The process of acting on the instructions contained in the active gene.
- **Transcriptomics**: The study of the total sum of mRNA which indicates gene expression levels.
- **Whole genome sequencing**: The impressive process of discovering the entire sequence of an organisms DNA in a single attempt, also known as high throughput sequencing.

QUEEN'S UNIVERSITY BELFAST

1. Global Genes. RARE Diseases: Facts and Statistics [Accessed 26/02/2018] Available from: https://globalgenes.org/rare-diseases-facts-statistics/
2. The 100,000 Genomes Project [Accessed 26/02/2018] Available from: https://www.genomicsengland.co.uk/the-100000-genomes-project/.

HUNTINGTON'S
DISEASE ASSOCIATION
NORTHERN IRELAND
Families at the heart of all we do

Support and fun for us designed by us

Sorcha McPhillips

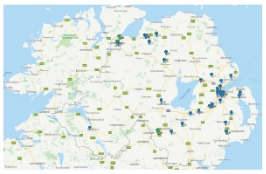
Huntington's Disease Association Northern Ireland

Support & Fun For Us Designed By Us

Empowering children & young adults to be their own HD hero

anxiety
parents
death future
responsibility
anger fear loneliness
worry testing
sadness changes
embarrassed
illness

Online surveys, focus groups, & parent engagement for maximum collaboration

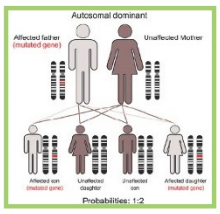


There are 94 children among the families HDANI supports across N. Ireland

HDANI youth

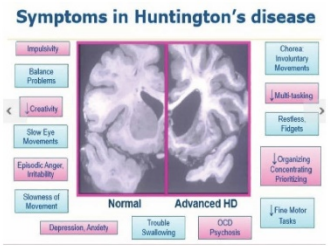
Play, art, music & animal therapy are used by our trained counsellor to encourage discussion about feelings & concerns, building resilience & confidence.

Allowing kids to be kids in a safe, fun, non judgmental space where they can form life-long friendships to help them tackle the road ahead



Youth committee works with staff & other children and young people to decide on venues, activities, branding, support themes, invited guests and consult on grant applications & project planning to ensure the service meets their needs

Ages 5-25 divided into age-appropriate groups for fun activities & group support



For more info email
youth@hdani.org.uk

Belfast Health and Social Care Trust

Inaugural rare-case simulation series: Improving rare condition education among NI Paediatric trainees

Peter Mallett[1], Ben McNaughten[2], Thomas Bourke[2], Andrew Thompson[2].

1. *Clinical Fellow in Education and Simulation. Royal Belfast Hospital for Sick Children (RBHSC) , Belfast Health & Social Care Trust*
2. *Department of Paediatric Education & Simulation, Royal Belfast Hospital for Sick Children*

Aims: The Royal College of Paediatrics and Child Health (RCPCH) provides a comprehensive curriculum for paediatric trainees. Achieving exposure to more uncommon paediatric conditions in clinical practice is often extremely challenging. We sought to address this by offering the regions paediatric trainees an opportunity to participate in new inter-professional, high-fidelity, simulated scenarios based on rare but important clinical conditions.

Methods: We surveyed all level three paediatric trainees (ST6-ST8) in our deanery enquiring about what conditions they had never experienced in clinical practice. We designed and embedded a new 'rare- case' simulation teaching programme into the postgraduate education schedule in the deanery's tertiary paediatric centre.

Results: Tailored simulation scenarios have been designed and currently run on a monthly regular basis through our 'rare case sunrise simulation sessions'. These include acute stroke, SVC obstruction and neonatal hyperammonaemia following trainee survey. Feedback demonstrated an improvement in confidence and clinical knowledge from the trainees following the sessions. Since inception in 2016, over 50 trainees have benefitted.

Conclusions: Rare-case simulation allows trainees to develop their confidence in managing uncommon conditions and address areas of the college curriculum which they have previously struggled to achieve. This has aided rare-disease awareness, promotion and improved paediatric trainee education in this area.

Inaugural rare-case simulation series: Improving rare condition education among NI Paediatric trainees

Dr.P Mallett[1], Dr B McNaughten[2], Dr C Hart[3], Dr T Bourke[4], Dr A Thompson[4]

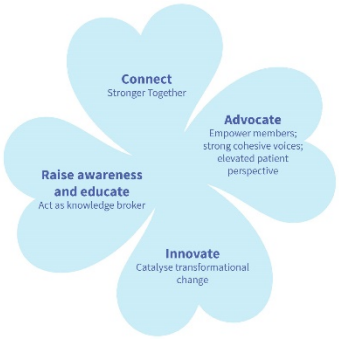
1-Clinical Fellow In Education & Simulation 2017-8 RBHSC,
2- Education Fellow 2016-17 RBHSC
3- Education Fellow 2015-2016 RBHSC,
4 -Department of Paediatrics & Child Health, RBHSC, Belfast Trust.

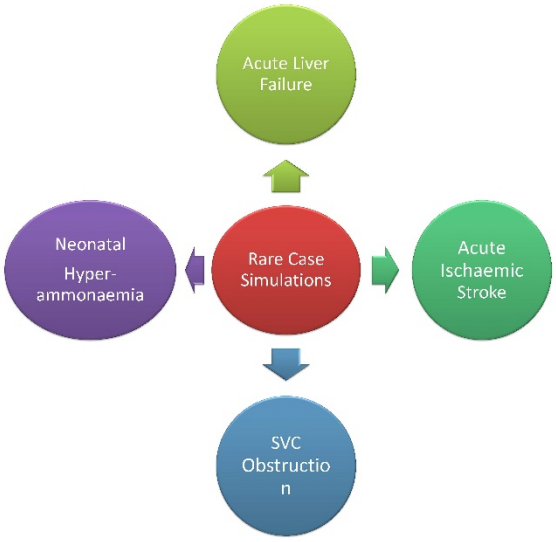
Aims & Objectives:

The Royal College of Paediatrics and Child Health (RCPCH) provides a comprehensive curriculum for paediatric trainees. Achieving exposure to more uncommon paediatric conditions in clinical practice is often extremely challenging. We sought to address this by offering the regions paediatric trainees an opportunity to participate in new inter-professional, high-fidelity, simulated scenarios based on rare but important clinical conditions.

Methodology:

We surveyed all level three paediatric trainees (ST6-ST8) in our deanery enquiring about what conditions they had never experienced in clinical practice. We designed and embedded a new 'rare- case' simulation teaching programme into the postgraduate education schedule in the deanery's tertiary paediatric centre in Royal Belfast Hospital for Sick Children.





Trainee Feedback:

" Rare case simulation training allows me to meet curriculum competencies which I have not previously been able to achieve."

" These sessions enabled me to develop my confidence in the management of conditions which I rarely see in clinical practice "

" Really useful simulation series. Provides us with chance to encounter more uncommon conditions in a safe learning environment. "

Results

Tailored simulation scenarios have been designed and currently run on a monthly regular basis through our 'rare case sunrise simulation sessions'.

These include acute stroke, SVC obstruction and neonatal hyperammonaemia following trainee survey. Feedback demonstrated an improvement in confidence and clinical knowledge from the trainees following the sessions.
Since inception in 2016, over 50 trainees have benefitted.

Conclusions:

Simulation-based training has been shown to be effective for trainees and has been widely used in many training programs.[2]

Rare-case simulation allows trainees to develop their confidence in managing uncommon conditions and address areas of the college curriculum which they have previously struggled to achieve. We plan to further extend our series with input from varying specialists and parents of patients with rare diseases.

This initiative has aided rare-disease awareness, promotion and improved paediatric trainee education in this area.

All-Island Rare Diseases Conference 2018

References:
1) Curriculum for Paediatric Training. General Paediatrics. Level 1, 2 and 3 Training. August 2016. http://www.rcpch.ac.uk/training-examinations-professional-development/postgraduate-training/
2) Barsuk JH, Cohen ER et al. Simulation-based education with mastery learning improves residents' LP skills. *Neurology*. 2012; 79(2):132–7.

@RBHSCSimteam

Peter.mallett@belfasttrust.hscni.net

AVERT – Autoimmunity Relapse Prediction using Multiple Parallel Data Sources

Julie Power

Vasculitis Ireland Awareness

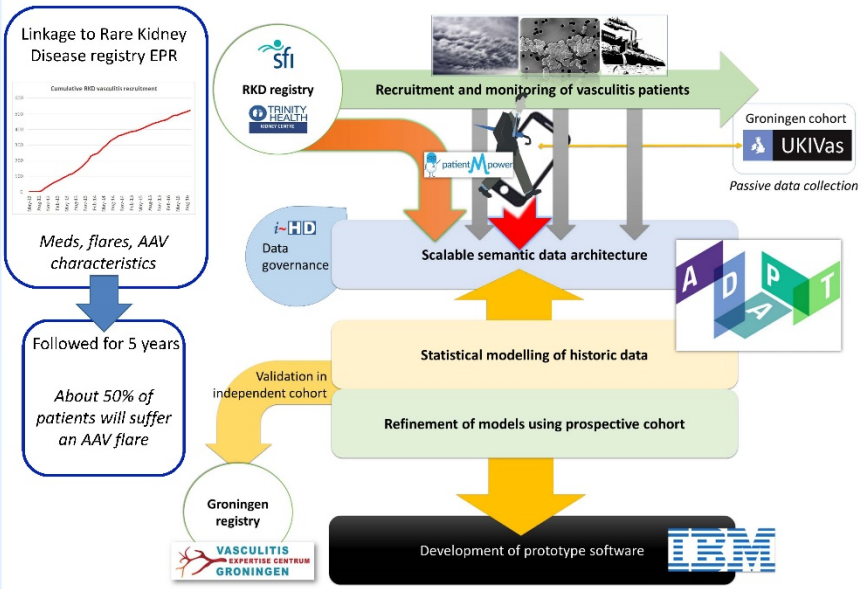
AVERT – Autoimmunity Relapse Prediction using Multiple Parallel Data Sources

Brian Reddy[1,2,3], Brett Houlding[3], Jennifer Scott[1], Lucy Hederman[2], Alan Meehan[2], Julie Power[4], Eamonn Costelloe[5], Martin Holmes[5], Daniel Dempsey[3], John Kelleher[2], Jason Wyse[3], Declan O'Sullivan[2], Mark A Little[1]

1. Trinity Health Kidney Centre, Trinity College Dublin; 2. ADAPT Centre for Digital Content; 3. School of Computer Science and Statistics, Trinity College Dublin; 4. Vasculitis Ireland Awareness; 5. patientMpower

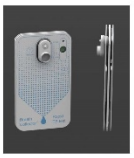
Background

Inaccurate prediction of disease flare exposes the 10% of the population with autoimmune disease to *overtreatment* with immune system suppressing drugs (with consequent infection and cancer risk), or *under-treatment* with these drugs (with consequent organ failure due to uncontrolled autoimmunity). **We need better tools to personalise these treatments.**
Using ANCA vasculitis as a model archetypal relapsing autoimmune disease, **we aim to identify and validate the environmental / clinical factor interactions influencing flare of the disease.**



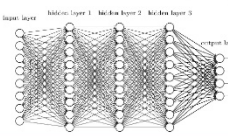
Breath condensate

Proteomic analysis of breath condensate is a novel technology that can provide a non-invasive estimate of systemic inflammation. We shall explore options for integrating this into the AVERT work flow



Statistical & Machine Learning analysis

Location
Time
Flare events
Medication
Phenotype
Weather
Pollution
Local infections



Iterative predictive algorithm building, refined with each new flare event

www.thkc.ie

Just how rare are rare lymphoid malignancies in Europe? Findings from RARECAREnet.

Charlene M. McShane[1] and Lesley A. Anderson[1] on behalf of the RARECARE working group.

[1]Centre for Public Health, Queen's University Belfast

Background: Rare cancers contribute to 22% of the total cancer burden in Europe and are associated with poorer survival when compared to more common cancers.

Methods: Using data from RARECAREnet, which collates data from 94 European population-based registries, we investigated incidence, prevalence and survival of rare lymphoid malignancies diagnosed in Europe.

Results: In 2008, an estimated n=931,855 individuals were living with a rare lymphoid malignancy in Europe. During 2001-2007, n=283,288 new rare lymphoid malignancies were diagnosed; age-adjusted incidence rate (ASR): 15.2/100,000 people. Higher incidence was observed in males (ASR 18.1 vs 12.8 per 100,000). Multiple myeloma/plasmacytoma (and heavy chain disease) and diffuse large B-cell lymphoma were the most commonly diagnosed malignancies accounting for 31.6% and 23.9% of cases respectively. Across all subtypes, slight increases in 5-year relative survival were observed between 1999 and 2007. Survival remains poor for prolymphocytic leukaemia B-cell (5-year relative survival: 31%), multiple myeloma/plasmacytoma (and heavy chain disease; 35%) and other T-cell lymphomas and Natural Killer cell neoplasms (38%).

Conclusions: Rare lymphoid malignancies are a significant public health problem. A collaborative international approach is required to advance research in this field and to ensure patients are afforded the same opportunities as patients diagnosed with common cancers.

[Poster withheld]

ART and family: Life, but not as you know it

Susie Rea

PhD Researcher, Research Institute of Art and Design, Ulster University

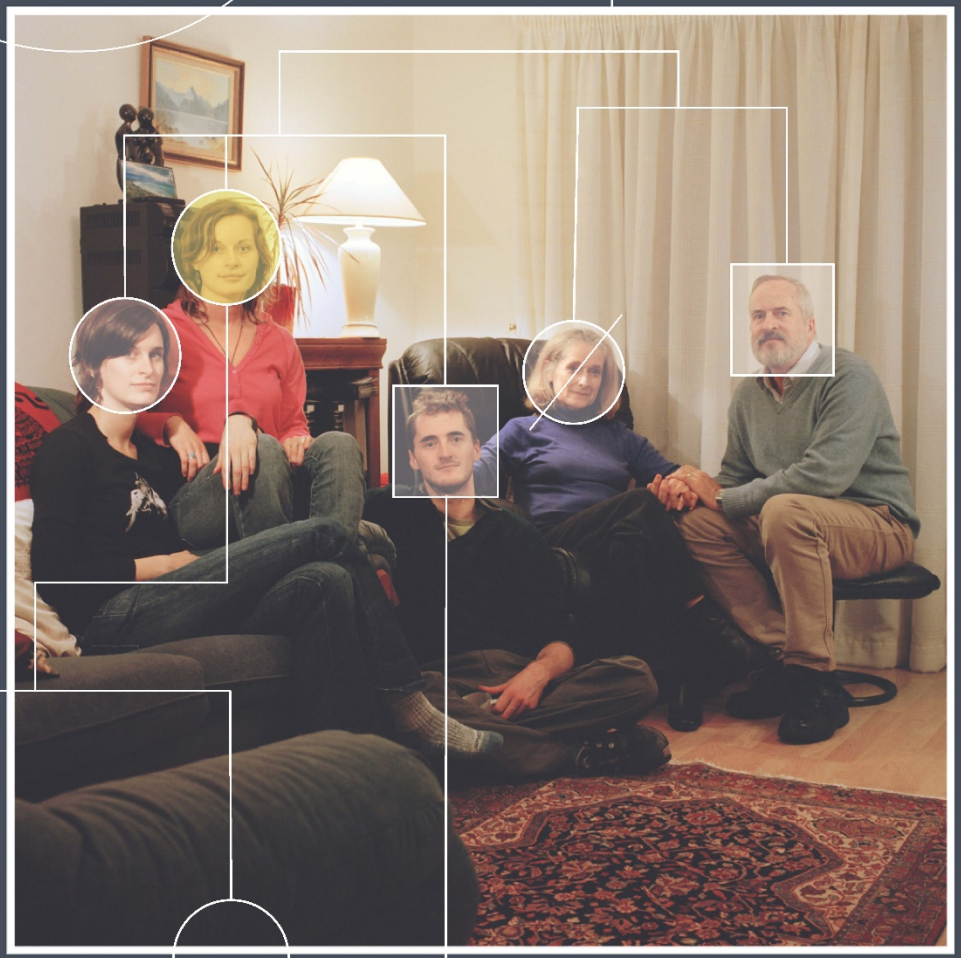
Genetics is about people. Research undertaken by biomedical scientists in support of the field touches people. We are the end users. However, the human experience of the application of these technologies is often side-lined. The work being produced by artists within this interdisciplinary field of study is no exception. Many practitioners have been preoccupied with life at a micro level and the technology that makes this molecular view possible. This trend in 'reductionism', though visually fascinating and conceptually interesting, has meant that we have lost sight of the "embodied individual" (Ayers, 2011) and the human experience. It is the purpose of this PhD research project to address this gap through the creation of a new body of work. The project looks at the experience of families living with genetic disease, with a specific focus on the choices they face as a result of developments in the area of advanced reproductive technologies (ART). If art can assist in communicating complex scientific, social and ethical ideas visually and experientially to a wider audience, it is the challenge of this artist to do so using the humble family photograph. Susie Rea is a practicing photographic artist and PhD researcher at Ulster University. Her work explores the fault lines where medicine, technology and society meet.

(ART) AND FAMILY:

LIFE, BUT NOT AS YOU KNOW IT

This project looks at the experience of families living with genetic disease, with a specific focus on the choices they now face as a result of advanced reproductive technologies (ART). If art can assist in communicating complex scientific, social and ethical ideas visually and experientially to a wider audience, the challenge of this project is to do so photographically, using the family portrait.

Susie Rea is a practicing artist and PhD researcher at Ulster University, Belfast. Her work explores the fault lines where medicine, technology and society meet.



Are we preoccupied with life at a molecular level? Have we lost sight of the 'embodied individual' and the human experience?

Can an artwork challenge the viewer to engage empathically?

ART presents ordinary families with extraordinary choices, choices which alter the genetic make-up of their family and that of society in general.